



## Overview

### Points To Remember About Osteopetrosis

- In osteopetrosis, bones become too dense but can easily break. This happens when new bone grows faster than old bone is removed.
- Bone that is too dense can cause serious problems throughout the body.
- Treatments for osteopetrosis depend on the type and how severe the disease is.
- Your doctor will advise you to practice healthy habits that include a balanced diet and proper dental care.

In osteopetrosis, bones are too dense but easily broken. Bone that is too dense can cause serious problems throughout the body. For example, pressure on nerves in the skull can cause vision loss, hearing loss, or paralysis of facial muscles. It can also cause crowding of the bone marrow (tissue inside the bones). This can lead to low levels of cells needed to fight infection, carry oxygen to the body's cells, or control bleeding.

### What happens in osteopetrosis?

Osteopetrosis occurs when bone development and breakdown are not balanced. This causes bones to be dense but easily broken. People are born with the condition.

### Who Gets

You need to have a gene from one or both parents to have the disorder.

# Types

Types of osteopetrosis include:

- The **malignant infantile form** is seen at or shortly after birth and can shorten life expectancy.
- The **adult form** is milder and may not be diagnosed until adolescence or adulthood. The formal name for this is autosomal dominant osteopetrosis.
- **Intermediate osteopetrosis** is found in children younger than age 10. This form is less severe than the malignant infantile form, but more severe than the adult form.

# Symptoms

The severity of symptoms differs for the three forms of osteopetrosis. The most common symptoms are:

- Bone fractures.
- Low blood cell levels.
- Impaired vision.
- Impaired hearing.
- Dental problems related to infection.

# Causes

Osteopetrosis is caused by defects in one or more genes involved in the formation, development, and function of cells that break down bone tissue during bone remodeling. Remodeling is a normal process in which old bone is removed and new bone is created to replace it.

The malignant infantile form occurs when a child inherits a copy of the defective gene from both parents. Only one copy of the defective gene is necessary for the adult form, so you can inherit this form from one parent. This can be passed from one parent. However, in many cases, the abnormal gene does not come from either parent, but may be the result of a new gene mutation that happens at conception.

# Diagnosis

Your doctor may do the following to test you for osteopetrosis and problems related to the disease:

- X-rays: Bones will appear unusually dense and chalky white.
- Hearing and vision tests.
- Blood tests.
- Other imaging scans, such as computerized axial tomography (CAT) scans and magnetic resonance imaging (MRI).

## Treatment

Treatment for osteopetrosis depends on the type and how severe the disease is. Treatments can include:

- Interferon gamma-1b, which is an injected drug designed to delay disease progression and the only therapy specifically approved by the U.S. Food and Drug Administration (FDA) for the disease.
- Calcitriol, the active form of vitamin D, can help reduce bone density.
- Prednisone, a hormone similar to cortisone produced by the body, may be taken in the short term to improve blood cell levels.
- Physical and occupational therapy to help children develop motor and other skills.
- Orthopaedic care for fractures.
- Regular doctor visits to monitor the eye, ear, nose, and throat.
- Bone marrow transplant: In children with the severe form of the disease. This can completely stop bone and bone marrow abnormalities, but it cannot reverse damage that has already occurred.

## Who Treats

Several different types of doctors may be involved in the diagnosis and treatment of osteopetrosis. Often patients see a team of doctors who work together to provide the best treatment. These doctors may include:

- Hematologists, who are blood disease specialists.
- Endocrinologists, who specialize in hormonal and metabolic disorders.
- Orthopaedists, who specialize in the treatment of, and surgery for, bone and joint diseases or injuries.
- Ophthalmologists, who specialize in eye care.
- Otolaryngologists, who are ear, nose, and throat (ENT) specialists.

# Living With

In addition to supervising your medical treatments, your doctor will advise you to practice healthy habits in your everyday life. These include:

- Eating a balanced diet to support normal growth and development.
- Practicing good dental care to decrease the chance of dental infections.

## Research Progress

Recent research into causes and treatments of osteopetrosis have focused on:

- Genetic and molecular mechanisms of abnormal bone formation and breakdown.
- Surgical management of complications.
- Modifications to bone marrow transplantation that would make the procedure more readily accessible to children shortly after diagnosis, with the hope of stopping the disease and preventing its complications.

## Related Resources

### **U.S. Food and Drug Administration**

Toll free: 888-INFO-FDA (888-463-6332)

Website: <https://www.fda.gov>

Drugs@FDA at <https://www.accessdata.fda.gov/scripts/cder/daf/>. Drugs@FDA is a searchable catalog of FDA-approved drug products.

### **Centers for Disease Control and Prevention, National Center for Health Statistics**

Website: <https://www.cdc.gov/nchs>

### **NIH Osteoporosis and Related Bone Diseases ~ National Resource Center**

Website: <https://www.bones.nih.gov>

### **Genetic and Rare Diseases Information Center**

Website: <https://rarediseases.info.nih.gov/GARD/>

### **Genetics Home Reference**

Website: <https://www.ghr.nlm.nih.gov/condition/osteopetrosis>

### **American Academy of Orthopaedic Surgeons**

Website: <https://www.aaos.org> (con información en español)

### **Orphanet**

Website: <https://www.orpha.net>

**St. Jude Children's Research Hospital**

Website: <https://www.stjude.org>

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at [NIAMSInfo@mail.nih.gov](mailto:NIAMSInfo@mail.nih.gov).

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